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ABSTRACT

This bibliography, prepared by the National Library of Medicine through a literature search of its online databases, covers all aspects of newborn screening. It includes references to screening for: inborn errors of metabolism, such as phenylketonuria and galactosemia; hemoglobinopathies, particularly sickle cell disease; congenital hypothyroidism and congenital adrenal hyperplasia; cystic fibrosis; and muscular dystrophy. Citations are primarily to journal articles from "MEDLINE," "HEALTH PLANNING AND ADMINISTRATION," and "BIOETHICSLINE." The bibliography contains over 750 references to English and foreign-language publications, issued between January 1980 and March 1987, in alphabetic order. (Author/JDD)

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LITERATURE SEARCH

NEWBORN SCREENING

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753 Citations

Prepared by Karen Patrias

Screening of infants in the first month of life for inherited metabolic disorders and other diseases requiring early diagnosis has been known for over thirty years, beginning with programs in the 1950's for detection of phenylketonuria (PKU). Although many advances have been made both in the numbers of disorders for which screening tests have been developed and in the sophistication and accuracy of the tests themselves, controversy currently exists over what constitutes an ideal newborn screening program. Specific issues include technical as well as socioeconomic questions such as what diseases should be included, which are the most effective testing methods, should screening programs be voluntary or mandatory and for what groups of infants, and what is their cost-benefit to society as a whole.

This bibliography covers all aspects of newborn screening and includes specific reference to screening for inborn errors of metabolism, such as PKU and galactosemia; hemoglobinopathies, particularly sickle cell disease; congenital hypothyroidism and congenital adrenal hyperplasia; cystic fibrosis; and muscular dystrophy. Citations are primarily to journal articles from NLM's MEDLINE, HEALTH PLANNING AND ADMINISTRATION, and BIOETHICSLINE databases.

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SEARCH STRATEGY

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SS 1 = INFANT, NEWBORN OR INFANT, NEWBORN, DISEASES/DIAGNOSIS

SS 2 = 1 AND MASS SCREENING OR 1 AND DIAGNOSTIC TESTS, ROUTINE

SS 3 = 1 AND ALL SCREEN:

SS 4 = EXP METABOLISM, INBORN ERRORS

SS 5 = EXP HYPOTHYROIDISM OR ADRENAL HYPERPLASIA, CONGENITAL

SS 6 = EXP HEMOGLOBINOPATHIES OR IGG OR ALL BIOTIN:

SS 7 = CYSTIC FIBROSIS OR MUSCULAR DYSTROPHY

SS 8 = 3 AND 4 OR 3 AND 5 OR 3 AND 6 OR 3 AND 7

SS 9 = (TW) ALL NEWBORN: OR ALL NEONAT:

SS10 = 9 AND ALL SCREEN: (TW)

SS11 = TS (TI) :SCREEN:

SS12 = 2 OR 8 OR 11

SS13 = 12 AND NOT ANIMAL

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